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Case report

Nail-patella syndrome with infertility in a 38 year old Saudi male: A case report

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Abstract

Nail-patella syndrome (NPS) which also known as hereditary osteo-onychodysplasia is a rare autosomal-dominant disorder characterized by a classic clinical tetrad of changes in the nails, knees, elbows and the presence of iliac horns.

Occasionally the central nervous system, the eyes and the renal system are affected.

NPS was reported worldwide, affecting males and females equally.

Infertility was not mentioned in this syndrome before.

Here we are reporting a case of NPS in a 38 year old Saudi male, in association with infertility, which was exceptional and could represent an additional manifestation to the syndrome.

Keywords: Nail patella syndromes; Infertility; Azoospermia; Sertoli cell-only syndrome

1. Introduction

Nail-patella syndrome (NPS), also known as hereditary osteoonychodysplasia (HOOD) or Turner-Kieser syndrome, is a rare syndrome with an incidence ranging from 4.5–22 per million. It is an autosomal dominant disorder. Both sexes are affected in equal numbers.

NPS is characterized by a clinical tetrad including fingernail dysplasia, hypoplastic or absent patellae, dislocation of the radial head, and bony protuberances of the iliac known as the iliac horns.

In addition, abnormalities such as renal dysplasia, muscle weakness, eye abnormality may be present.

No cases of NPS associated with infertility were reported in the literature. We report a case with NPS and infertility.

2. Case history

A 38 year old Saudi male presented to our O.P.D With a history of total finger nail dystrophy since birth, knee deformity, pain and instability, and a limitation of elbow joint movement. He also gave a history of palmoplantar hyperhidrosis.

He is a product of non-consanguineous marriage, with similar illness in the family (mother, brother).

Abbreviations: NPS, nail patella syndrome; SCOS, sertoli cell-only syndrome.

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He is divorced. He has been married for more than 2 years with no siblings; his wife was medically free with normal fertility.

His present physical examination shows: total nail dystrophy affecting all his fingers, with a loss of the creases in the skin overlying the all-distal interphalangeal joints (Fig. 1(a) and (b)).

Musculoskeletal examination: shows underdevelopment of arms and legs proximal musculature, cubital fossa webbing, hyper extensible knee joints and pes planus (flat foot) (Fig. 2(a), (b)).

Complete blood count, renal and liver function tests were normal. There was mild protein urea detect by 24 h urine collection.

X-rays show hypo plastic patella bilaterally and posterior iliac horns (Fig. 3).

Our patient was also diagnosed to have infertility by the urology department due to the inability to achieve pregnancy after one year of unprotected intercourse.

His wife's evaluation for infertility turned out to be normal.

There was no history of decreased libido, galactorrhea or erectile dysfunction.

The patient has no history of testicular trauma, STD, urogenital infections, radiation, hormonal treatment or medication exposure, mumps, chronic systemic disease, tuberculosis or hypothyroidism.

He exhibited normal sexual character without gynecomastia. There was no goiter.

The testes were normal in size, with a normal shape and consistency, no varicocele. The rest of the systemic examination was normal.

His Testosterone and Luteinizing hormones were in normal levels, but the follicular stimulation hormone was elevated three times the reference range.

Semen analysis shows azoospermia.

Ultra-sonogram of the scrotum showed normal testicular volume with normally visualized epididymis, vas deferens and there was no evidence of varicocele.

The testicular biopsy revealed complete absence of all germ cell layers in the seminiferous tubules, with sertoli cells only.

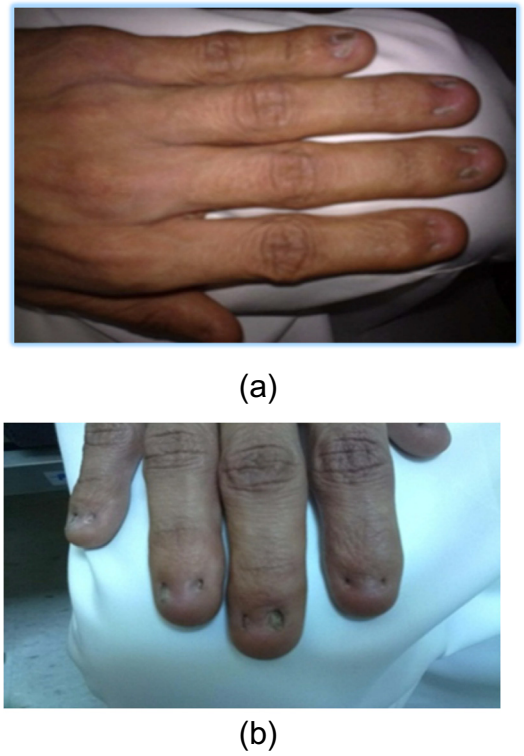

Fig. 1. (a) and (b) Shows total nail dystrophy.

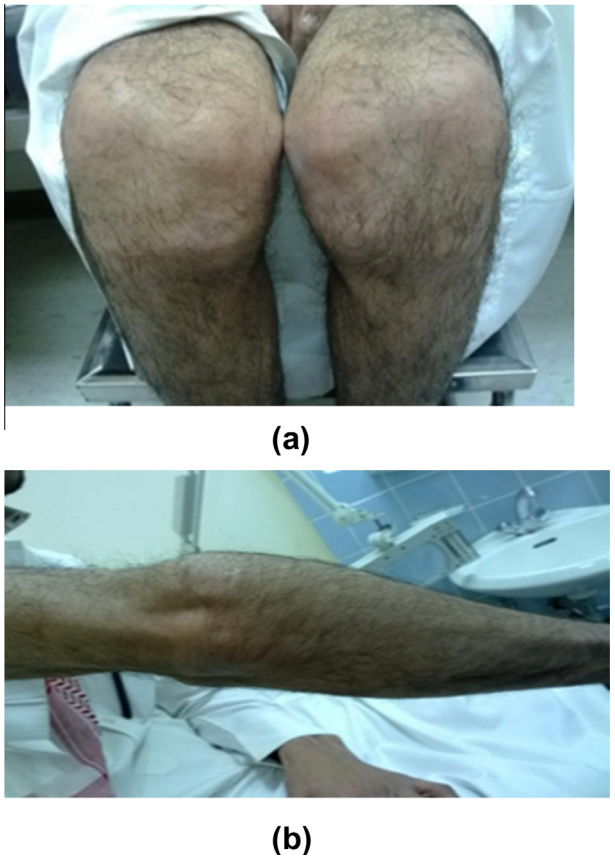

Fig. 2. (a) and (b) Shows abnormal knee and elbow shape due to absent patella and webbing of the antecubital fossa respectively.

3. Discussion

The NPS was recognized more than 70 years ago; as an inherited syndrome but only recently the responsible gene identified, which is the LMX1B gene.

LMX1B gene was located at the distal end of the long arm of chromosome 9. It is a transcription factor of the LIM-homeodomain type that plays an important role for limb and renal development in vertebrates.

More than 140 heterozygous mutations in LMX1B have been reported, including missense, splicing, deletions, and nonsense mutations. Most mutations are located in the LIM domains. LMX1B gene mutations are fully penetrant,

Fig. 3. Absent patella on knee X-ray.

but there is variability of expression even within families (Álvarez-Martín et al., 2013).

To diagnose NPS, nail dysplasia and patellar hypoplasia are essential findings.

Other diagnostic features are hypoplasia of the radial head and iliac prominences (which are known as iliac horns) (Dreyer et al., 1998).

Renal complications and ophthalmologic problems can accompany nail-patella syndrome.

Nail and distal digital changes that are observed in NPS include Nail hypoplasia, dystrophic changes (discoloration, ridging and splitting), Triangular lunulae, that is pathognomonic finding in 80–90% of the cases and loss of the creases in the skin overlying the distal interphalangeal joint (Nandedkar-Thomas and Scher, 2005).

3.1. Bone and joint involvement

Includes knee deformity and pain due to Patellae dysplasia (ranges from complete agenesis to hypoplasia) with a tendency to dislocate laterally.

Elbow Dysplasia, the radial head may be small and sub laxed dorsally, leading to impairment of forearm movement, and webbing of the antecubital fossa.

The iliac horns are one of the most characteristic but harmless features of the NPS (Bongers et al., 2002).

3.2. Renal disease

Proteinuria due to glomerulopathy and end-stage renal disease, which is relatively rare and is reported in approximately 5% of patients (Kamath and Bhagwandas, 2010).

3.3. Other anomalies in NPS include

Deformities of the sternum, spina bifida occulta, clinodactyly of the fifth fingers, muscle weakness, impaired hearing, waddling gate, and scapular winging, hyperhidrosis, hypothyroidism and goiters, mental retardation, keratoconus, microcornea, microphakia, and cataracts (Bongers et al., 2002; Kamath and Bhagwandas, 2010) but infertility was not reported before.

Infertility, a common condition with important psychological, and medical implications, is defined as the inability to achieve conception despite one year of frequent unprotected intercourse.

In general males with infertility have either oligozoospermia (decrease in number of sperm cells in the ejaculate compared to reference ranges) or azoospermia (no sperm cells in the ejaculate).

The causes of male infertility can be divided into four main areas

- Hypothalamic pituitary.
- Testicular disease.
- Post-testicular defects.
- Idiopathic.

Our patient has a testicular defect; there was an absence of the testicular germ cells.

Primary testicular failure occurs in approximately 1% of all males. It is known as sertoli-cell-only syndrome (SCOS) or germinal cell aplasia.

Most causes of SCOS syndrome are idiopathic.

These patients exhibit normal secondary male sexual characteristics, semen analysis shows azoospermia or severe oligozoospermia, high FSH and with normal testosterone levels, but ultimately a diagnosis of SCOS depends on histo-pathology which reveals absence of germ cells. There is no known effective medical therapy available (Hanmayyagari et al., 2015).

4. Conclusion

To our knowledge, primary idiopathic infertility was not reported in NPS and it could be an additional feature to the syndrome.

Conflict of interest

None.

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